

## CLAIMS

1. An isolated nucleic acid comprising any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof.

5 2. An isolated nucleic acid comprising at least eight consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof.

3. An isolated nucleic acid comprising at least 80% nucleotide identity with a nucleic acid comprising any one of SEQ ID NOs: 1-4, or a complementary nucleotide  
10 sequence thereof.

4. The isolated nucleic acid according to claim 3, wherein the nucleic acid comprises an 85%, 90%, 95%, or 98% nucleotide identity with the nucleic acid comprising any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof.

5. An isolated nucleic acid that hybridizes under high stringency conditions  
15 with a nucleic acid comprising any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof.

6. An isolated nucleic acid comprising a nucleotide sequence as depicted in any one of SEQ ID NOs: 1-4, or of a complementary nucleotide sequence thereof.

7. A nucleotide probe or primer specific for the ABCA12 gene, wherein the  
20 nucleotide probe or primer comprises at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or of a complementary nucleotide sequence thereof.

8. A nucleotide probe or primer specific for the ABCA12 gene, wherein the nucleotide probe or primer comprises a nucleotide sequence of any one of SEQ ID NO: 7-  
25 38, or a complementary nucleotide sequence thereof.

9. The nucleotide probe or primer according to any of claim 7 or 8, wherein the nucleotide probe or primer comprises a marker compound.

10. A method of amplifying a region of the nucleic acid according to claim 1, wherein the method comprises:

30 a) contacting the nucleic acid with two nucleotide primers, wherein the first nucleotide primer hybridizes at a position 5' of the region of the nucleic acid, and the

second nucleotide primer hybridizes at a position 3' of the region of the nucleic acid, in the presence of reagents necessary for an amplification reaction; and

b) detecting the amplified nucleic acid region.

11. A method of amplifying a region of the nucleic acid according to claim 10,  
5 wherein the two nucleotide primers are selected from the group consisting of

a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or of a complementary nucleotide sequence,

b) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID  
10 NOs: 7-38, or a complementary sequence thereof.

12. A kit for amplifying the nucleic acid according to claim 1, wherein the kit comprises:

a) two nucleotide primers whose hybridization position is located respectively 5' and 3' of the region of the nucleic acid; and optionally,

15 b) reagents necessary for an amplification reaction.

13. The kit according to claim 12, wherein the two nucleotide primers are selected from the group consisting of

a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or of a complementary nucleotide  
20 sequence,

b) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOs: 7-38, or a complementary sequence thereof.

14. A method of detecting a nucleic acid according to claim 1, wherein the method comprises:

25 a) contacting the nucleic acid with a nucleotide probe selected from the group consisting of

1) a nucleotide probe comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof,

30 2) a nucleotide probe as in any one of claims 7-9,

3) a nucleotide probe comprising a nucleotide sequence of any one of SEQ ID NOs: 7-38, or a complementary nucleotide sequence thereof, and

b) detecting a complex formed between the nucleic acid and the probe.

15. The method of detection according to claim 14, wherein the probe is immobilized on a support.

16. A kit for detecting the nucleic acid according to claim 1, wherein the kit  
5 comprises

a) a nucleotide probe selected from the group consisting of 1) a nucleotide probe comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOs: 1-4, or a complementary nucleotide sequence thereof, 2) a nucleotide primer as in any one of claim 7 or 9, 3) a nucleotide probe  
10 comprising a nucleotide sequence of any one of SEQ ID NOs: 7-38, or a complementary nucleotide sequence thereof, and optionally,

b) reagents necessary for a hybridization reaction.

17. The kit according to claim 16, wherein the probe is immobilized on a support.

18. A recombinant vector comprising the nucleic acid according claim 1.

19. The vector according to claim 18, wherein the vector is an adenovirus.

20. A recombinant host cell comprising the recombinant vector according to claim 19.

21. A recombinant host cell comprising the nucleic acid according claim 1.

22. An isolated nucleic acid encoding a polypeptide comprising an amino acid sequence of any one of SEQ ID NO: 5 or 6.

23. A recombinant vector comprising the nucleic acid according to claim 22.

24. A recombinant host cell comprising the nucleic acid according to claim 22.

25. A recombinant host cell comprising the recombinant vector according to claim 23.

26. An isolated polypeptide selected from the group consisting of

a) a polypeptide comprising an amino acid sequence of any one of SEQ ID NOs: 5 or 6,

b) a polypeptide fragment or variant of a polypeptide comprising an amino acid sequence of any one of SEQ ID NOs: 5 or 6, and

c) a polypeptide homologous to a polypeptide comprising amino acid sequence of any one of SEQ ID NO: 5 or 6.

27. An antibody directed against the isolated polypeptide according to claim 26.

28. The antibody according to claim 27, wherein the antibody comprises a detectable compound.

29. A method of detecting a polypeptide, wherein the method comprises

- 5 a) contacting the polypeptide with an antibody according to claim 28; and  
b) detecting an antigen/antibody complex formed between the polypeptide and the antibody.

30. A diagnostic kit for detecting a polypeptide, wherein the kit comprises

- a) the antibody according to claim 28; and  
10 b) a reagent allowing detection of an antigen/antibody complex formed between the polypeptide and the antibody.

31. A pharmaceutical composition comprising the nucleic acid according to claim 1 and a physiologically compatible excipient.

32. A pharmaceutical composition comprising the recombinant vector according  
15 to claim 23 and a physiologically compatible excipient.

33. Use of a recombinant vector according to claim 18 for the manufacture of a medicament for the prevention and/or treatment of a subject affected by a dysfunction in the lipophilic substance transport.

34. Use of an isolated ABCA12 polypeptide comprising an amino acid sequence  
20 of SEQ ID NO: 5 or 6 for the manufacture of a medicament intended for the prevention and/or treatment of a subject affected by a dysfunction in the lipophilic substance transport or by a pathology located on the chromosome locus 2q34 such as for example the lamellar ichthyosis, the polymorphic congenital cataract, or insulin-dependant diabete mellitus.

35. A pharmaceutical composition comprising a polypeptide comprising an  
25 amino acid sequence of any one of SEQ ID NOs: 5 or 6, and a physiologically compatible excipient.

36. Use of an ABCA12 polypeptide comprising an amino acid sequence of any  
one of SEQ ID NOs: 5 or 6 for screening an active ingredient for the prevention or  
treatment of a disease resulting from a dysfunction in the lipophilic substance transport or of  
30 a pathology located on the chromosome locus 2q34 such as for example the lamellar ichthyosis, the polymorphic congenital cataract, or insulin-dependant diabete mellitus.

37. Use of a recombinant host cell expressing an ABCA12 polypeptide comprising an amino acid sequence of any one of SEQ ID NOs: 5 or 6, for screening an active ingredient for the prevention or treatment of a disease resulting from a dysfunction in the lipophilic substance transport.

5 38. A method of screening a compound active on the transport of lipid substance, an agonist, or an antagonist of ABCA12 polypeptides, wherein the method comprises

a) preparing a membrane vesicle comprising ABCA12 polypeptide having SEQ ID NOs: 4 or 5 and a lipid substrate comprising a detectable marker;

10 b) incubating the vesicle obtained in step a) with an agonist or antagonist candidate compound;

c) qualitatively and/or quantitatively measuring a release of the lipid substrate comprising the detectable marker; and

15 d) comparing the release of the lipid substrate measured in step b) with a measurement of a release of a labeled lipid substrate by a membrane vesicle that has not been previously incubated with the agonist or antagonist candidate compound.

39. A method of screening an agonist or an antagonist of ABCA12 polypeptides, wherein the method comprises

20 a) incubating a cell that expresses at least a ABCA12 polypeptide having SEQ ID NOs: 4 or 5 with an anion labeled with a detectable marker;

b) washing the cell of step a) whereby excess labeled anion that has not penetrated into the cell is removed;

c) incubating the cell obtained in step b) with an agonist or antagonist candidate compound for the ABCA12 polypeptide;

25 d) measuring efflux of the labeled anion from the cell; and

e) comparing the efflux of the labeled anion determined in step d) with efflux of a labeled anion measured with a cell that has not been previously incubated with the agonist or antagonist candidate compound.

40. An implant comprising the recombinant host cell according to claim 24.